

Abstract

Methods for detecting in a single assay any one of multiple chromosomal disorders that result from aneuploidy or certain mutations, particularly microdeletions, and kits for use therein. A polymerase chain reaction (PCR) is carried out to amplify eukaryotic genomic DNA using a plurality of primer oligonucleotide pairs wherein one primer of each pair has a detectable label attached 5' thereto. A plurality of the primer pairs are targeted to DNA segments of different chromosomes of interest which are indicative of potential chromosomal disorders, and one pair is targeted for a control gene. The amplified PCR products are purified, and single-stranded DNA having the detectable labels is obtained therefrom and hybridized with spots on a microarray that each contain DNA oligonucleotide probes having nucleotide sequences complementary to a nucleotide sequence of one strand of each segment. The microarray is imaged for presence of labels on its respective spots, and the absence or presence of chromosomal disorders as indicated by one or more of the targeted DNA segments of interest is diagnosed by first comparing the imaging results to the imaging of spots specific to the control gene and then to results obtained from imaging normal DNA.